Not an Established Condition

- 3 - Not an Established Condition

3-Methylcrotonyl-Coenzyme A (CoA) Carboxylase Deficiency

33-34 weeks EGA (if the birth weight is >1325 grams or 2 lbs 15 oz)

3rd Nerve Palsy (aka: Third Nerve Palsy; Third Cranial Nerve Palsy; 3rd Cranial Nerve Palsy; Oculomotor Palsy; Oculomotor Nerve Palsy; Cranial Nerve III Palsy)

- 6 - Not an Established Condition

6th Nerve Palsy (aka: Abducens palsy; Lateral rectus palsy; Cranial mononeuropathy VI)

- A - Not an Established Condition

Aarskog-Scott syndrome

Abnormal Neurological Exam at Discharge from NICU

Absent Septum Pellucidum (aka: Absence Septum Pellucidum, Absent Cavum Septum Pellucidum)

Achondroplasia

Acute Lymphoid Leukemia (aka: ALL)

Adams Oliver Syndrome (aka: AOS; Absence Defect of Limbs, Scalp, and Skull; Congenital Scalp Defects with Distal Limb Reduction Anomalies; Aplasia Cutis Congenita with Terminal Transverse Limb Defects)

Adjustment Disorder (as defined within DC:0-3R, and diagnosed by specially-qualified professional)

Alagille Syndrome (aka: AHD; Arteriohepatic Dysplasia; Cholestasis with Peripheral Pulmonary Stenosis; Syndromatic Hepatic Ductular Hypoplasia)

Albinism (aka Ocular Cutaneous Albinism, Oculocutaneous Albinism, Ocular Albinism)

Alcohol-Related Neurodevelopmental Disorder (aka: ARND; Fetal Alcohol Spectrum Disorder; FASD)

Alport Syndrome (aka: Hematuria-Nephropathy Deafness; Hemorrhagic Familial Nephritis; Hereditary Deafness and Nephropathy; Hereditary Nephritis With Sensory Deafness; Hereditary Nephritis and Nerve Deafness)

Alstrom Syndrome (aka: ALMS)

Amniotic Band Syndrome (affecting fetus or newborns)

Aniridia

Anophthalmia, unilateral

Anoxic Insult to Brain (aka anoxic brain injury)

Anterior Jaw Displacement (aka: mandibular enlargement; prognathism)

Anterior segment dysgenesis

Anxiety Disorder Not Otherwise Specified (NOS) (as defined within DC:0-3R, and diagnosed by specially-qualified professional)

APGAR score of five or less at five minutes

Arachnoid Cyst

Arnold-Chiari Malformation (AKA: Chiari 1 Malformation; Chiari Malformation)

Auditory Neuropathy (aka: Auditory neuropathy/auditory dyssynchrony; Auditory neuropathy/auditory dys-synchrony; AN/AD)

Aural Atresia, Bilateral or Unilateral (aka: congenital aural atresia; CAA)

- B - Not an Established Condition

Beals Syndrome (aka: Beals-Hecht Syndrome; Arachnodactyly, Contractural Beals Type; Contractural Arachnodactyly, Congenital; CCA)

Beckwith Wiedemann Syndrome (aka: BWS; Beckwith-Syndrome; Exomphalos-Macroglossia-Gigantism Syndrome; EMG Syndrome; Hypoglycemia with Macroglossia; Macroglossia-Omphalocele-Visceromegaly Syndrome; Omphalocele-Visceromegaly-Macroglossia Syndrome; Visceromegaly-Umbilical Hernia-Macroglossia Syndrome; Wiedmann-Beckwith Syndrome)

Benign Enlargement of the Subarachnoid Space in infancy (aka: BESS)

Biliary Atresia (note: biliary agenesis and liver transplant)

Bjornstad Syndrome (aka: Deafness and Pili Torti, Bjornstad Type; Pili Torti and Nerve Deafness; Pili Torti-Sensorineural Hearing Loss)

Blue Diaper Syndrome (aka: Drummond's Syndrome: Hypercalcemia, Familial, with Nephrocalcinosis and Indicanuria)

Bowen Hutterite Syndrome (aka:Bowen-Conradi Hutterite Syndrome, Bowen-Conradi Syndrome, Hutterite Syndrome, Bowen-Conradi Type)

Brachial Plexus Palsy (aka: Duchenne's Paralysis, Duchenne-Erb Paralysis, Duchenne-Erb Syndrome, Erb's Paralysis, Erb-Duchenne Palsy, Erb-Duchenne Paralysis, Upper Brachial Plexus Palsy, Erb-Duchenne Type, or Upper Brachial Plexus Paralysis, Erb-Duchenne Type; Brachial Plexus Injury)

Branchio-oculo-facial syndrome (aka:BOFS, Branchiooculofacial Syndrome, Hemangiomatous Branchial Clefts-Lip Pseudocleft Syndrome, Imperforate Nasolacrimal Duct, and Premature Aging Syndrome, Lip Pseudocleft-hemangiomatous Branchial Cyst Syndrome)

Bronchopulmonary Dysplasia (BPD)

- C - Not an Established Condition

Carboxylase Deficiency, Multiple (aka: Biotinidase Deficiency, Carboxylase Deficiency, Multiple, Holocarboxylase Synthetase Deficiency, MCD)

Carpenter Syndrome (aka: ACPS II, Acrocephalopolysyndactyly Type II)

Cartilage hair hypoplasia syndrome (aka: CHH; Metaphyseal chondrodysplasia, McKusick type)

Cataracts, Congenital

Catel-Manzke Syndrome (aka: Catel-Manzke Type Palatodigital Syndrome, Hyperphalangy-Clinodactyly of Index Finger with Pierre Robin Syndrome, Index Finger Anomaly with Pierre Robin Syndrome, Pierre Robin Syndrome with Hyperphalangy and Clinodactyly)

Caudal Regression Syndrome (aka: Caudal Dysplasia; Caudal Dysplasia Sequence; Sacral Agenesis, Congenital; Sacral Regression)

Cerebral Bleed (aka: intracerebral bleed, cerebral hemorrhage, intracerebral hemorrhage; brain hemorrhage; parenchymal brain hemorrhage; intraparenchymal hemorrhage; intraparenchymal hematoma; periventricular hemorrhage)

Cerebrocostomandibular Syndrome (aka: CCM Syndrome, CCMS, Rib Gap Defects with Micrognathia)

Chandler's Syndrome (aka: Dystrophia Endothelialis Cornea, Iris Atrophy with Corneal Edema and Glaucoma)

Choanal Atresia (unilateral or bilateral)

Choroid Plexus Cyst

Chromosome 17q12 Deletion

Chromosome 5q minus syndrome

Chronic Ear Infections

Citrullinemia (aka: Citrullinuria)

Cleft Lip

Cleft Palate

Cleft Palate and Cleft Lip

Clinodactyly

Clonus (aka Clonospasm)

Club Foot (includes bilateral)

Cogan Syndrome

Cogan-Reese Syndrome (aka: ICE Syndrome, Cogan-Reese Type; Iridocorneal Endothelial (ICE) Syndrome, Cogan-Resse Type; Iris Naevus Syndrome; Iris Nevus Syndrome)

Coloboma

Complex congenital heart disease (aka: heterotaxy (left-right reversal of heart and other internal organs), complete AVSD (atrioventricular septal defect), multiple VSDs (ventricular septal defect), Swiss cheese type septum (holes in the septum of the heart), PHTN (pulmonary hypertension), Tetralogy of Fallot / TOF)

Cone Dystrophy (aka: Combined Cone-Rod Degeneration; Cone-Rod Degeneration; Cone-Rod Degeneration, Progressive; Cone-Rod Dystrophy; Retinal Cone Degeneration; Retinal Cone Dystrophy; Retinal Cone-Rod Dystrophy)

Congenital Adrenal Hyperplasia (aka: CAH)

Congenital hypopituitarism and congenital disc pigmentation syndrome

Congenital Nystagmus

Congenital skeletal dysplasia. (aka: Skeletal Dysplasia)

Congenital Syphilis

Congenital Toxoplasmosis

Congestive Heart Failure (aka: CHF; Heart Failure)

Connexin 30 gene mutation

Conradi-Hunermann syndrome (aka: Chondrodysplasia Punctata, X-linked Dominant Type; Chondrodystrophia Calcificans Congenita; Conradi Disease; Dysplasia Epiphysialis Punctata)

Craniosynostosis without Radial Defects

Crigler-Najjar Syndrome (aka: Bilirubin Glucuronosyltransferase Deficiency Type I; Congenital Familial Nonhemolytic Jaundice Type I; Uridine Diphosphate Glucuronosyltransferase, Severe Def. Type I)

Crouzon Syndrome (aka: Craniofacial Dysotosis; Craniostenosis (Crouzon Type); Crouzon Craniofacial Dysostosis)

Cystic Fibrosis

- D - Not an Established Condition

Depression of Infant and Early Childhood: Type II - Depressive Disorder NOS (as defined within DC:0-3R, and diagnosed by specially-qualified professional)

Diaphragmatic Hernia

Diastematomyelia

Disorders of Relating and Communicating (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted)

Drug Exposure, Prenatal (aka: neonatal abstinence syndrome)

Duane Syndrome

Dysphagia (aka: Dysphagia Oropharyngeal)

- E - Not an Established Condition

Eagle-Barrett syndrome (aka: Prune Belly Syndrome)

Eales Disease (aka: Eales Retinopathy, Idiopathic Peripheral Periphlebitis)

Ear, Patella, Short Stature Syndrome (aka: EPS; Meier-Gorlin Syndrome; Microtia, Absent Patellae, Micrognathia Syndrome)

Ectrodactyly Ectodermal Dysplasia Cleft Lip/Palate (aka: EEC Syndrome; Ectrodactyly-Ectodermal Dysplasia-Clefting Syndrome)

Ehlers Danlos Syndrome (aka: E-D Syndrome; ED Syndrome)

Ellis Van Creveld Syndrome (aka: Chondroectodermal Dysplasia; Mesoectodermal Dysplasia)

Empty Sella Syndrome (aka: Empty Sella Turcica)

Encephalits, Japanese (aka: JE, Japanese B Encephalitis, Russian Autumnal Encephalitis, Summer Encephalitis)

Epidermolysis Bullosa

Epidermolytic Hyperkeratosis

Esophogeal Atresia

Exostoses, Multiple

Extraocular Fibrosis Syndrome (aka: Congenital Fibrosis of the Extraocular Muscles; CFEOM)

- F - Not an Established Condition

Fabry Disease (aka:Alpha-Galactosidase A Deficiency; Anderson-Fabry Disease; Angiokeratoma Corporis Diffusum; Angiokeratoma Diffuse; Ceramide Trihexosidase Deficiency; GLA Deficiency; Hereditary Dystopic Lipidosis)

Facial Palsy (aka: Bell's Palsy; Facial Nerve Palsy; 7th Nerve Palsy)

Facioscapulohumeral Muscular Dystrophy (aka: FMD; FSH; FSHD; Facio-Scapulo-Humeral Dystrophy; Muscular Dystrophy, Facioscapulohumeral; Muscular Dystrophy, Landouzy Dejerine)

Failure to Thrive

Familial Dysautonomia (aka: Riley-Day Syndrome; FD; Hereditary Sensory and Autonomic Neuropathy, Type III (HSAN, Type III); Hereditary Sensory Neuropathy Type III; HSAN III; HSN III)

Familial exudative vitreoretinopathy (aka: FEVR)

Fanconi Syndrome (aka: de Toni-Fanconi syndrome)

Feeding Disorder, Sensory Food Aversions (as defined within DC:0-3R, and diagnosed by specially-qualified professional)

Femoral Facial Syndrome (aka: Femoral Dysgenesis, Bilateral; Femoral Dysgenesis, Bilateral-Robin Anomaly; Femoral Hypoplasia-Unusual Facies Syndrome)

Fetal Alcohol Spectrum Disorder (aka: Alcohol-related neurodevelopmental disorder; alcohol-related neurobehavioral disorder; fetal alcohol effects)

Fetal Valproate Syndrome (aka: Dalpro, (Fetal Effects From); Depakene, (Fetal Effects From); Depakote Sprinkle (Fetal Effects From); Depakote, (Fetal Effects From); Divalproex, (Fetal Effects From); Epival (Fetal Effects From); Fetal Anti-Convulsive Syndrome; Myproic Acid (Fetal Effects From); Valproic Acid (Fetal Effects From))

Fibromatosis, Congenital Generalized (aka: CGF; Infantile Myofibromatosis; IM)

Forbes Disease (aka: Amylo-1,6-Glucosidase Deficiency; Cori Disease; Debrancher Deficiency; Glycogen Storage Disease III; Glycogenosis Type III; Limit Dextrinosis)

Fracture of Vertebral Column with Spinal Cord Injury

Fraser Syndrome (aka: Cryptophthalmos Syndrome; Cryptophthalmos-Syndactyly Syndrome; Cyclopism Fraser-Francois Syndrome; Meyer-Schwickerath's Syndrome; Ulrich-Feichtiger Syndrome)

Freeman Sheldon Syndrome (aka: Craniocarpotarsal dystrophy (dysplasia); DA2A; Distal arthrogryposis type 2A; FSS; Whistling face syndrome; Whistling face-windmill vane hand syndrome)

Frontofacionasal Dysplasia (aka: Cleft Lip-Palate, Blepharophimosis, Lagophthalmos, and Hypertelorism; Facio-Fronto-Nasal Dysplasia; Frontofacionasal Dysostosis; Nasal-fronto-faciodysplasia)

Frontonasal Dysplasia (aka: FND; Median Cleft Face Syndrome)

- G - Not an Established Condition

Galloway-Mowat Syndrome (aka: Galloway Syndrome; Hiatal Hernia-Microcephaly-Nephrosis, Galloway Type; Microcephaly-Hiatal Hernia-Nephrosis, Galloway Type; Microcephaly-Hiatal Hernia-Nephrotic Syndrome; Nephrosis-Microcephaly Syndrome; Nephrosis-Neuronal Dysmigration Syndrome)

Gastroschisis (aka: Abdominal Wall Defect; Aparoschisis)

Gaucher Disease, Type I (aka: Sphingolipidosis 1)

Glutaricaciduria I (aka: Dicarboxylic Aminoaciduria; GA I; Glutaric Acidemia I; Glutaric Aciduria I; Glutaricacidemia I; Glutaryl-CoA Dehydrogenase Deficiency; Glutaurate-Aspartate Transport Defect)

Glutaricaciduria II (aka: Electron Transfer Flavoprotein, Deficiency of; Electron Transfer Flavoprotein: Ubiquinone Oxidoreductase, Deficiency of; GA II; Glutaric Acidemia II; Glutaric Aciduria II; Glutaricacidemia II; MADD; Multiple Acyl-Co-A Dehydrogenation Deficiency)

Goldenhar Syndrome (aka: Oculo-Auriculo-Vertebral/OAV syndrome)

Goltz Syndrome (aka: Gorlin Syndrome; nevoid basal cell carcinoma syndrome)

Goodman Syndrome (aka: ACPS IV; Acrocephalopolysyndactyly Type IV)

Gordon Syndrome (not the other "Gordon Syndrome with hypertension, hyperkalaemia, and normal glomerular filtration rate") (aka: Arthrogryposis Multiplex Congenita, Distal, Type IIA; Camptodactyly-Cleft Palate-Clubfoot; Distal Arthrogryposis, Type IIA)

Gorlin-Chaudhry-Moss Syndrome (aka: Craniofacial Dysostosis-PD Arteriosus-Hypertrichosis-Hypoplasia of Labia; Craniosynostosis-Hypertrichosis-Facial and Other Anomalies; GCM Syndrome)

Gottron Syndrome (aka: H. Gottron's syndrome; acrogeria, Gottron type; familial acrogeria; familial acromicria)

Greig Cephalopolysyndactyly Syndrome (aka: Frontodigital Syndrome (obsolete); GCPS; Hootnick-Holmes Syndrome (obsolete); Polysyndactyly with Peculiar Skull Shape; Polysyndactyly-Dysmorphic Craniofacies, Greig Type)

Griscelli Syndrome, type 2

Griscelli Syndrome, type 3

Guillain Barre Syndrome (aka: Acute Autoimmune Peripheral Neuropathy; Acute Immune-Mediated Polyneuropathy ; Acute Inflammatory Demyelinating Polyneuropathy; Acute Inflammatory Demyelinating Polyradiculoneuropathy; Acute Inflammatory Neuropathy; Acute Inflammatory Polyneuropathy; GBS; Landry's Ascending Paralysis; Landry-Guillain-Barre-Strohl Syndrome; Post-Infective Polyneuritis)

- **H - Not an Established Condition**

Hajdu Cheney Syndrome (aka: HCS)

Hallermann Streiff syndrome

Hand-Schuller-Christian (aka: Histiocytosis X; Hemophagocytic Lymphohistiocytosis, Familial, 1)

Hanhart Syndrome (aka: aglossia-adactylia; hypoglossia-hypodactylia syndrome; peromelia with micrognathia)

Hartnup Disease

Hay-Wells Syndrome

Hearing Loss (diagnosis with unilateral permanent hearing loss)

Hemihyperplasia (aka: hemihypertrophy)

Hemophilia

Hepatoblastoma

Hereditary Hyperphosphatasia

Hermansky Pudlak Syndrome

Herpes-Zoster (aka: Hunt Syndrome)

Heterotopia (aka Subependymal Nodular Heterotopia)

Holt Oram Syndrome (aka: HOS)

Horner's Syndrome (aka: Horner Syndrome)

Huntington's Disease

Hydrops (aka: hydrops fetalis; erythroblastosis fetalis)

Hyperbilirubinemia (aka: Jaundice of the newborn; Neonatal hyperbilirubinemia)

Hyperexplexia

Hypertonia

Hypochondroplasia

Hypophosphatasia

Hypophosphatemia, Familial

Hypoplastic Left Heart Syndrome

Hypothyroidism, Congenital

Hypotonia, Congenital, Non-Benign Form

- I - Not an Established Condition

Ichthyosis Congenita (aka: Ichthyosis Lamellar; Autosomal Recessive Congenital Ichthyosis, Ichthyosis, Harlequin Type)

Ichthyosis, Chanarin Dorfman Syndrome

Infantile Botulism

Intestinal malrotation (aka: malrotation of the intestine)

Intraventricular Hemorrhage (aka: IVH, periventricular hemorrhage, PVH) Grade II

Intraventricular Hemorrhage (IVH) Grade I (aka: Germinal Matrix Hemorrhage; Grade I IVH; Grade I periventricular hemorrhage, Grade I PVH)

IPEX Syndrome (aka: immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome)

IRF6-Related Disorders

Isovaleric Acidemia

Ivemark Syndrome (aka: Asplenia Syndrome, Bilateral Right-Sidedness Sequence, Splenic Agenesis Syndrome)

- J - Not an Established Condition

Jackson-Weiss Syndrome

Jansen Type Metaphyseal Chondrodysplasia

Jarcho-Levin Syndrome

Jejunal atresia

Job Syndrome

Juvenile Myelomonocytic Leukemia

Juvenile Rheumatoid Arthritis (aka: JRA)

- K - Not an Established Condition

Kartagener Syndrome

Kawasaki Disease

Kearns Sayre Syndrome

Kenny-Caffey Syndrome

Keratosis Follicularis

Klippel-Feil Syndrome (aka: Klippel-Feil Sequence)

Klippel-Trenaunay Syndrome (aka: Klippel-Trenaunay-Weber Syndrome)

Kniest Dysplasia

- L - Not an Established Condition

Laband Syndrome

LADD Syndrome

Langerhans Cell Histiocytosis

Laron Syndrome

Larsen Syndrome

Laryngomalacia

Laryngotracheoesophageal cleft (aka: LTEC)

Lead level >10 ug/dL

Legg Calve Perthes Disease

LEOPARD Syndrome

Leprechaunism

Leri Pleonosteosis

Leukocytosis

Loeys-Dietz Syndrome

Loken Senior Syndrome

Lumbosacral Agenesis

Lyme Disease

Lymphadenopathy

- M - Not an Established Condition

Macrocephaly

Macroglossia

Macular lesion (of the eye) (aka: lesion of the macula)

Maffucci Syndrome

Marcus Gunn Phenomenon

Marfan Syndrome

Maroteaux Lamy Syndrome

Marshall Syndrome

Mastocytosis

Maxillonasal Dysplasia, Binder Type

McCune Albright Syndrome

McKusick Type Metaphyseal Chondrodysplasia

Megacystis Microcolon Intestinal Hypoperistalsis Syndrome (aka: Berdon Syndrome; Megacystis Microcolon Intestinal Hydronephrosis Syndrome; Neonatal Hollow visceral myopathy)

Meige Syndrome

Melkersson Rosenthal Syndrome

Melnick Needles Syndrome

Melorheostosis

Metabolic Acidosis

Metaphyseal Chondrodysplasia, Schmid Type

Metatarsus adductus (aka: Metatarsus varus; Forefoot varus)

Metatropic Dysplasia I

Methylmalonic Aciduria without Glycinemia, Group 5 (aka: Methylmalonic Acidemia; ketotic hyperglycinemia)

Micrognathia

Micromelia (aka: nanomelia)

Microphthalmia

Microtia

Microtia and Atresia (aka: Microtia/Atresia; Microtia and Aural Atresia; Anotia)

Microvillus Inclusion Disease

Miller Syndrome (aka: Postaxial Acrofacial Dysostosis Syndrome)

Missing Fingers (aka: missing thumbs; absent fingers; absent thumbs)

Mixed Disorder of Emotional Expressiveness (as defined within DC:0-3R, and diagnosed by specially-qualified professional)

Morquio Syndrome

Motonia Congenita - Thomsen Disease

Moyamoya Syndrome

Mulibrey Nanism Syndrome (Perheentupa Syndrome)

Mulvihill Smith Syndrome

MURCS Association

Muscular Dystrophies, Limb Girdle

Muscular Dystrophy, Becker

Muscular Dystrophy, Emery Dreifuss

Muscular Dystrophy, Oculo Gastrointestinal

Myoclonus, General

Myositis Ossificans Progressiva

Myotonia Congenita

- N - Not an Established Condition

Nager Syndrome

Nail Patella Syndrome

Necrotizing Enterocolitis (NEC) (aka: short gut/bowel syndrome)

Neonatal abstinence syndrome (NAS) is a term for a group of problems a baby experiences when withdrawing from exposure to narcotics.

Neonatal Hemochromatosis (NH)

Neonatal Herpes Simplex (HSV)

Neonatal Seizures (aka: Seizures of Newborn)

Nephrotic Syndrome (Aka: Membranous glomerulonephritis; Membranoproliferative glomerulonephritis; Mesangiocapillary glomerulonephritis)

Neuroblastoma

Neuromyotonia

Neutropenia

Nezelof Syndrome

NF1-Neurofibromatosis (aka: NF-1; NF1; Von Recklinghausen Disease)

NF2-Bilateral Acoustic Neurofibromatosis (aka Neurofibromatosis Type 2 (NF-2))

Night-Waking Disorder (Night-Waking Protodyssomnia) (as defined within DC:0-3R, and diagnosed by specially-qualified professional)

Noonan Syndrome

- O - Not an Established Condition

Ochoa Syndrome

Ocular Motor Apraxia, Cogan Type (aka: Congenital Oculomotor Apraxia; oculomotor apraxia, Cogan type; COMA; saccade initiation failure, congenital)

Oculo-Dento-Digital Dysplasia (aka: Dento-Oculo-Osseous Dysplasia ; ODD Syndrome ; ODDD ; ODOD ; Oculo Dento Digital Dysplasia ; Oculo-Dento-Osseous Dysplasia ; Oculodentodigital Dysplasia ; Osseous-Oculo-Dento Dysplasia)

Ollier Disease (aka: Dyschondroplasia; Enchondromatosis; Multiple Cartilaginous Enchondroses; Multiple Enchondromatosis)

Omphalocele

Optic atrophy (aka: optic nerve atrophy)

Organ Failure (other than brain/CNS), including but not limited to renal/kidney failure, hepatic/liver failure (aka: end-organ failure; multiple organ failure; mulitple system organ failure; multiple organ system failure; multiple organ dysfunction syndrome (MODS))

Orotic Aciduria

OSMED, Heterozygous (aka: Arthro-Ophthalmopathy; Epiphyseal Changes and High Myopia; Ophthalmoarthropathy Weissenbacher-Zweymuller Syndrome; Hereditary Arthro-Ophthalmopathy; Stickler syndrome type I; Stickler syndrome type II)

OSMED, Homozygous (aka: oto-spondylo-megaepiphyseal dysplasia, autosomal recessive; oto-spondylo-megaepiphyseal dysplasia, homozygous; Nance-Sweeney syndrome)

Osteodystrophy, Congenital

Osteogenesis Imperfecta

- P - Not an Established Condition

Pachydermoperiostosis

Paget's Disease

Pallister Hall Syndrome

Panhypopituitarism

Panhypopituitarism (aka: Panhypopit; Combined Pituitary Hormone Deficiency)

Papillitis

Papillon Lefevre Syndrome

Paramyotonia Congenita

Paraneoplastic Neurologic Syndromes

Parry Romberg Syndrome

Pars Planitis

Parsonage Turner Syndrome

Pediatric Cardiomyopathy

Pentalogy of Cantrell

PEPCK Deficiency

Peters Anomoly (aka: bilateral anterior segment dysgenesis)

Peutz Jeghers Syndrome

Pfeiffer Syndrome Type I

PHACES Syndrome (aka: PHACE syndrome; Posterior fossa malformations, Hemangiomas, Arterial anomalies, Coarctation of aorta/Cardiac abnormalities, Eye anomalies, and Sternal defects)

Pharyngeal Dysphagia

Pica

Pierre-Robin Sequence (aka Perre-Robin Syndrome)

PIGA (aka: phosphatidylinositol glycan anchor biosynthesis class A gene mutation)

Pineal Cysts, Symptomatic

Plagiocephaly

Pleuropulmonary Blastoma

POEMS Syndrome

Poland Syndrome

Polychondritis

Polycystic Kidney Disease

Polydactyly (all types: postaxial, preaxial, and central)

Polymicrogyria, unilateral focal

Polymyalgia Rheumatica

Polymyositis

Porphyria, Acute Intermittent

Porphyria, Congenital Erythropoietic

Porphyria, Hereditary Coproporphyria

Porphyria, Variegate

Positive Toxicology Screen

Posterior Uveitis

Potter Sequence (aka: Potter Syndrome; Oligohydramnios)

Primary alveolar hypoventilation (aka Ondine's Curse)

Progeria, Hutchinson Gilford

Progressive Osseous Heteroplasia (POH)

Prolonged Bereavement/Grief Reaction (as defined within DC:0-3R, and diagnosed by specially-qualified professional)

Proteus Syndrome

Prune Belly Syndrome (aka Eagle-Barrett syndrome)

Pseudo-achondroplasia (aka: Pseudoachondroplastic Dysplasia)

Pseudohypoparathyroidism

Pseudoxanthoma Elasticum (PXE)

PTEN Hamartoma Tumor Syndrome

Pterygium Syndrome, Multiple

Ptosis

Pulmonary Eventration (aka: Eventration of the Diaphragm)

Pyknodysostosis

Pyloric stenosis

Pyridoxine-Dependent Seizures (PDS)

Pyruvate Kinase Deficiency

- R - Not an Established Condition

Rabson-Mendenhall Syndrome

Ramsay-Hunt Syndrome

Rapp Hodgkin Syndrome (aka: Ectodermal dysplasia, Rapp-Hodgkin type; Ectodermal dysplasia, anhidrotic, with cleft lip and cleft palate; RHS; Rapp-Hodgkin (hypohidrotic)

ectodermal dysplasia syndrome)

Reflex Sympathetic Dystrophy Syndrome (aka: Algodystrophy ; Algoneurodystrophy ; Causalgia Syndrome (Major) ; Complex Regional Pain Syndrome; RSDS ; Reflex Neurovascular Dystrophy ; Sudeck's Atrophy)

Regulation Disorders of Sensory Processing: Hypersensitive, Type B: Negative / Defiant (as defined within DC:0-3R, and diagnosed by specially-qualified professional)

Respiratory Distress Syndrome, Infant (aka: Hyaline Membrane Disease; IRDS; Infantile Respiratory Distress Syndrome)

Retinal Hemorrhage

Retinitis Pigmentosa (aka: Progressive Pigmentary Retinopathy and RP)

Retinoblastoma

Retinopathy of Prematurity (aka: ROP and Retrolental Fibroplasia (obsolete))

Retinopathy, Arteriosclerotic (aka: Arteriosclerosis, Retina)

Retinopathy, Diabetic

Retinopathy, Hypertensive

Retinoschisis (aka: Congenital Retinal Cyst ; Congenital Vascular Veils in the Retina ; Giant Cyst of the Retina; Vitreoretinal Dystrophy) ((Disorder Subdivisions: Blessig Cysts; Congenital Retinoschisis; Familial Foveal Retinoschisis; Iwanoff Cysts; Peripheral Cystoid Degeneration of the Retina; Retinoschisis, Juvenile; Retinoschisis, Senile; Retinoschisis, Typical)

Reye Syndrome (aka: Fatty Liver with Encephalopathy; RS; Reye's Syndrome)

Rh incompatibility

Rieger Syndrome (aka: Axenfeld-Rieger Syndrome; Goniodysgenesis-Hypodontia; Iridogoniodysgenesis With Somatic Anomalies; RGS)

Robinow Syndrome (aka: Acral Dysostosis with Facial and Genital Abnormalities; Costovertebral segmentation defect with mesomelia (formerly); Fetal Face Syndrome; Robinow Dwarfism)

Romano Ward Syndrome (aka: Autosomal Dominant Long QT Syndrome; LQTS1; Long QT Syndrome Type 1; Long QT Syndrome without Deafness; RWS; Romano-Ward Long QT Syndrome; Ward-Romano Syndrome)

Rosai-Dorfman Disease (RDD; SHML; Sinus Histiocytosis with Massive Lymphadenopathy)

Roseola Infantum (aka: Exanthem Subitum; Pseudorubella; Sixth Disease)

Rothmund Thomson Syndrome (aka: Poikiloderma Atrophicans and Cataract; Poikiloderma Congenitale; RTS)

Russell Silver Syndrome (aka: RSS; Russell Syndrome; Russell-Silver Dwarfism; SRS; Silver Syndrome; Silver-Russell Dwarfism; Silver-Russell Syndrome)

- S - Not an Established Condition

Saethre Chotzen Syndrome

Sakati Syndrome

Sandifer Syndrome

Schinzel Syndrome

Scleroderma

Scoliosis

Seizures

Septo-optic Dysplasia (aka: Optic Nerve Hypoplasia (ONH); Pituitary Hypoplasia)

Setleis Syndrome

Severe Combined Immunodeficiency (aka: SCID; Severe immune deficiency)

Short Chain Acyl CoA Dehydrogenase Deficiency (SCAD)

Shwachman Syndrome

Simian B Virus Infection

Sjogren Syndrome

Sleep Behavior Disorder (as defined within DC:0-3R, and diagnosed by specially-qualified professional)

Sleep-Onset Disorder (Sleep-Onset Progodyssomnia) (as defined within DC:0-3R, and diagnosed by specially-qualified professional)

Sneddon Syndrome

Spinal Cord Injury (aka: SCI) (due to the wide range of severity and outcomes, functional impairment will be used to determine eligibility)

Split Hand/Split Foot Malformation

Spondyloepiphyseal Dysplasia Tarda

Sprengel Deformity

Stenosis, Spinal

Stevens Johnson Syndrome

Stickler Syndrome

Stuve-Wiedemann Syndrome

Subdural Hematoma

Substance Exposed Newborn (aka: Prenatal Substance Exposure; Prenatal Drug Exposure; Drug-Exposed Infant; Drug-Exposed Newborn)

Summitt Syndrome

Syndactyly (aka: polysyndactyly; webbing of the fingers and/or toes)

Syringobulbia

Systemic Herpes Simplex Virus (HSV) Infection (aka: disseminated herpes simplex infection)

- T - Not an Established Condition

Telecanthus with Associated Abnormalities

Tethered Spinal Cord Syndrome

Tetrahydrobiopterin Deficiency (including the following subdivisions: Tetrahydrobiopterin Synthesis, GTP Cyclohydrolase I (GTPCH) Deficiency, 6-Pyruvoyl Tetrahydropterin Synthase (PTPS) Deficiency [aka: Hyperphenylalanemia with Biopterin Defect, PTPS Deficiency], Tetrahydrobiopterin Regeneration, Pterin-4-alpha-Carbinolamine Dehydratase (PCD) Deficiency, Dihydropteridine Reductase (DHPR) Deficiency)

Tetralogy of Fallot

Thalamic Syndrome (Dejerine Roussy)

Three M Syndrome

Thrombocytopenia

Tietze Syndrome

Tolosa Hunt Syndrome

TORCH Syndrome

Torticollis

Tourette Syndrome

Townes Brocks Syndrome

Toxic Epidermal Necrolysis

Toxic Optic Neuropathy (aka: Toxic/Nutritional Optic Neuropathy)

Tracheoesophageal Fistula (aka, Esophageal Atresia and Tracheoesophageal Fistula; Esophageal Fistula)

Tracheomalacia

Tracheostomy (aka: trach; trach tube; tracheostomy tube)

Treacher Collins Syndrome

Tricho Dento Osseous Syndrome

Trichorhinophalangeal Syndrome Type I

Trichorhinophalangeal Syndrome Type III

Trigger Finger

Trismus Pseudocamptodactyly Syndrome

Truncus Arteriosus

Turcot Syndrome

Twin-Twin Transfusion Syndrome (aka: TTTS)

Tyrosinemia, Hereditary

- U - Not an Established Condition

Unilateral Blindness

- V - Not an Established Condition

VACTERL Association

Valinemia (aka: Hypervalinemia; Valine Transaminase Deficiency)

Vascular Malformations of the Brain (aka: Cerebral Malformations, Vascular; Intracranial Vascular Malformations; Occult Intracranial Vascular Malformations)

Vasculitis (aka: angiitis)

Vasculitis, Cutaneous Necrotizing (aka: CNV; Cutaneous Leukocytoclastic Angiitis; Dermal Necrotizing Angiitis; Hypersensitivity Vasculitis)

Ventricular Septal Defects (aka: Hole in the Heart, VSD, Congenital Ventricular Defects)

Ventriculomegaly

Vertebral anomalies (aka: fusion of vertebrae; hemi-vertebra; hemivertebra)

Very Long Chain Acyl CoA Dehydrogenase Deficiency (aka: VLCAD; ACADL; LCAD)

Vocal Cord Nodules

Vocal Cord Paresis (aka: vocal cord paralysis), whether unilateral (left or right) or bilateral

Vogt Koyanagi Harada Syndrome (aka: Alopecia-Poliosis-Uveitis-Vitiligo-Deafness-Cutaneous-Uveo-Oto Syndrome; Harada Syndrome; Uveomeningitis Syndrome; VKH Syndrome)

Von Hippel-Lindau Syndrome (aka: Angiomatosis Retinae ; Lindau Syndrome; Pheochromocytoma; Renal cell carcioma; Retinocerebellar Angiomatosis; VHL; VHL Syndrome; Von Hippel Lindau disease)

- W - Not an Established Condition

Waardenburg Syndrome (aka: Waardenburg Syndrome Type I (WS1); Waardenburg Syndrome Type II (WS2); Waardenburg Syndrome Type IIA (WS2A); Waardenburg

Syndrome Type IIB (WS2B); Waardenburg Syndrome Type III (WS3) ; Waardenburg Syndrome Type IV (WS4))

Waldmann Disease

Weismann Netter Stuhl Syndrome

Whipple Disease

Wildervanck Syndrome (aka cervicooculoacoustic syndrome)

Wilm's Tumor

Winchester Syndrome

Wolff-Parkinson-White Syndrome (aka: WPW)

Wyburn Mason Syndrome

- **X - Not an Established Condition**

X linked Lymphoproliferative Syndrome (aka: XLP)

- **Y - Not an Established Condition**

Yunis Varon Syndrome

Established Condition

- 1 - Established Condition

10q25 deletion syndrome

10q26 deletion syndrome

1p36 deletion Syndrome (aka: Chromosome 1p36 deletion)

- 2 - Established Condition

22q11.2 Deletion Syndrome (aka: DiGeorge syndrome; velocardiofacial syndrome; Deletion 22q11.2 syndrome; Shprintzen syndrome; 22q deletion syndrome)

22q13 deletion syndrome (aka: Phelan-McDermid Syndrome)

- A - Established Condition

Achondrogenesis I (Parenti-Fraccaro)

Achondrogenesis II (Langer-Saldino)

Acrocallosal Syndrome, Schinzel Type (aka: Schinzel Acrocallosal Syndrome; Acrocallosal Syndrome, Schinzel Type; ACLS; ACS; Hallux duplication, postaxial polydactyly, absence of the corpus callosum, severe mental retardation syndrome)

Acrodysostosis (aka: Arkless-Graham; Acrodysplasia; Maroteaux-Malamut; Peripheral Dysostosis-Nasal Hypoplasia-Mental Retardation Syndrome; PNM Syndrome)

Adrenoleukodystrophy (aka: ALD; AMN; X-ALD; Addison disease and cerebral sclerosis; Adrenomyeloneuropathy; Siemerling-creutzfeldt disease; Bronze schilder disease; Schilder Disease; Melanodermic Leukodystrophy; sudanophilic leukodystrophy; Pelizaeus Merzbacher Brain Sclerosis; Pelizaeus-Merzbacher Disease; PMD; Sclerosis, Diffuse Familial Brain; Sudanophilic Leukodystrophy, Pelizaeus-Merzbacher Type)

Agenesis of the Corpus Callosum (aka: Absence of the corpus callosum; Hypogenesis of the corpus callosum)

Aicardi Syndrome (aka: Agenesis of Corpus Callosum and Chorioretinal Abnormality; Agenesis of Corpus Callosum With Chorioretinitis Abnormality; Agenesis of Corpus Callosum With Infantile Spasms And Ocular Anomalies; Chorioretinal Anomalies with Agenesis of the Corpus Callosum; Callosal Agenesis and Ocular Abnormalities)

Alexander Disease

Allan Herndon Syndrome (aka: Allan-Herndon-Dudley Syndrome (AHDS); Allan-Herndon-Dudley Mental Retardation)

Alper Disease (aka: Alper's Diffuse Degeneration of Cerebral Gray Matter with Hepatic Cirrhosis; Alpers Progressive Infantile Poliodystrophy; Christensen's disease; Christensen-Krabbe disease; Diffuse Cerebral Degeneration in Infancy; Poliodystrophia Cerebri Progressiva; Progressive Cerebral Poliodystrophy)

Amputation of the leg at the hip

Anencephaly

Angelman Syndrome (aka: AS)

Aniridia Cerebellar Ataxia Mental Deficiency (aka: Gillespie syndrome; Aniridia, Partial-Cerebellar Ataxia-Oligophrenia; Aniridia-Cerebellar Ataxia-Mental Retardation)hrenia)

Anophthalmia, bilateral

Apert Syndrome (aka: ACS 1; ACS I; Acrocephalosyndactyly, Type I; Syndactylic Oxycephaly)

APGAR score of 3 or less at 20 minutes

Aphasia (with evidence of brain damage) (aka: aphemia with evidence of brain damage)

Argininosuccinic aciduria (aka: Arginino Succinase Deficiency; ASA Deficiency; Argininosuccinate Lyase Deficiency; ASL Deficiency)

Arthrogryposis (aka: arthrogryposis multiplex congenita; AMC; multiple congenital contractures)

Asphyxia (with evidence of brain damage within athe first couple days of the event)

Ataxia Telangiectasia (aka: AT; Cerebello-Oculocutaneous Telangiectasia; Immunodeficiency with Ataxia Telangiectasia)

Autism Spectrum Disorder (aka: Infantile Autism; Kanner Syndrome; Autisitc Disorder; Asperger Syndrome; Asperger's Disorder; Pervasive Developmental Disorder; PDD; Pervasive Developmental Disorder-Not Otherwise Specified; PDD-NOS)

- B - Established Condition

Baller Gerold Syndrome (aka: Craniosynostosis with Radial Defects, Craniosynostosis-Radial Aplasia Syndrome)

Bannayan Riley Ruvalcaba (aka: Bannayan Riley Ruvalcaba Syndrome; BRRS, Bannayan-Zonana syndrome (BZS); Macrocephaly, multiple lipomas, and hemangiomata; Macrocephaly, pseudopapilledema, and multiple hemangiomata; Riley-Smith syndrome; Ruvalcaba-Myhre-Smith syndrome; RMSS; Ruvalcaba Syndrome)

Bardet-Biedl Syndrome (aka: Biedl-Bardet Syndrome; Laurence Moon-Biedl)

Bartter's syndrome (aka: Hypokalemic Alkalosis with Hypercalciuria)

Batten Disease (aka: CLN3, JNCL, Neuronal Ceroid Lipofuscinosis, Juvenile, Spielmeyer-Sjogren Disease, Vogt-Spielmeyer Disease, or Vogt-Spielmeyer-Sjogren Disease)

Borjeson Syndrome (aka: BORJ; Borjeson-Forssman-Lehmann Syndrome; BFLS)

Brain Tumor (aka: brain cancer; brain teratoma; glioma; astrocytoma; glioblastoma multiforme; ependymoma; oligodendroglioma; medulloblastoma; meningioma;

Schwannoma; acoustic neuroma; craniopharyngioma; germ cell tumor of the brain; germinoma; pineal region tumor; frontal lobe tumor)

- C - Established Condition

C Syndrome (aka: Opitz Trigonocephaly Syndrome, Trigonocephaly "C" Syndrome, Trigonocephaly Syndrome)

Cardiofaciocutaneous syndrome (aka: CFC syndrome, Cardio-facial-cutaneous syndrome, Facio-cardio-cutaneous syndrome)

Cerebellar Agenesis (aka: Cerebellar Aplasia; Cerebellar Hemiagenesis; Cerebellar Hypoplasia; Cerebellar Atrophy)

Cerebral atrophy

Cerebral Dysgenesis

Cerebral Palsy (aka: Ataxia Cerebral Palsy,Athetoid Cerebral Palsy, Congenital Cerebral Palsy, Diplegia of Cerebral Palsy,Hemiparesis of Cerebral Palsy, Hemiplegia of Cerebral Palsy, Hemiplegia, Hemiparesis, Postnatal Cerebral Palsy, Quadriparesis, Quadriparesis of Cerebral Palsy, Quadriplegia of Cerebral Palsy, Spastic Cerebral Palsy, Tetraparesis)

Cerebro Oculo Facio Skeletal Syndrome (aka: COFS Syndrome, Cerebrooculofacioskeletal Syndrome, Cockayne Syndrome type II, Pena Shokeir II Syndrome, Pena Shokeir Syndrome Type II)

Cerebromalacia (aka: Encephalomalacia)

CHARGE Syndrome (aka: CHARGE Association)

Chromosome 10, Monosomy 10p (aka: 10p Deletion Syndrome (Partial); Chromosome 10, 10p- Partial; Chromosome 10, Partial Deletion (short arm))

Chromosome 11, Partial Monosomy 11q (aka: 11q- Syndrome; Partial, Deletion 11q Syndrome, Partial; Distal 11q Monosomy; Distal 11q- Syndrome; JBS; Jacobsen Syndrome; Monosomy 11q, Partial; Partial Monosomy of Long Arm of Chromosome 11)

Chromosome 11, Partial Trisomy 11q (aka: 11q Partial Trisomy, Chromosome 11; Partial Trisomy 11q13-qter, Chromosome 11; Partial Trisomy 11q21-qter;, Chromosome 11; Partial Trisomy 11q23-qter; Distal Trisomy 11q; Partial Trisomy 11q; Trisomy 11q, Partial)

Chromosome 12 deletion

Chromosome 12p duplication

Chromosome 13, Partial Monosomy 13q (aka: 13q- Syndrome, Partial; Deletion 13q Syndrome, Partial; Monosomy 13q, Partial; Partial Monosomy of the Long Arm of Chromosome 13)

Chromosome 14 deletion

Chromosome 14 Ring (aka: Ring 14; Ring Chromosome 14; r14)

Chromosome 14, Trisomy Mosaic (aka: Trisomy 14 Mosaic; Trisomy 14 Mosaicism Syndrome; Trisomy 14 Syndrome)

Chromosome 15 Ring (aka: Ring 15; Ring 15 Chromosome; Ring 15 Chromosome (mosaic pattern); r15)

Chromosome 15, Distal Trisomy 15q (aka: Chromosome 15, Trisomy 15q2; Distal Duplication 15q; Partial Duplication 15q Syndrome)

Chromosome 15q11-q13 duplication (aka: Isodicentric 15; Inverted duplication 15)

Chromosome 16 Duplication

Chromosome 16, partial monosome 16p (aka: Partial Deletion of Chromosome 16p; 16p11.2 deletion syndrome)

Chromosome 16, partial monosomy 16q (aka: Partial deletion of Chromosome 16q)

Chromosome 17p13 duplication (aka: 17p13.3 microduplication syndrome; 17p13.2 duplication)

Chromosome 17p13.1 and/or 17p13.2 microdeletion

Chromosome 17q12 Duplication

Chromosome 18 Ring (aka: Ring 18; Ring Chromosome 18; r18)

Chromosome 18, Tetrasomy 18p (aka: Tetrasomy, Short Arm of Chromosome 18)

Chromosome 19p duplicaton

Chromosome 20q Trisomy (aka: Chromosome 20q Duplication; Partial Trisomy 20q; Trisomy 20q11.2; Chromsome 20q11.2 Duplication; Trisomy 20)

Chromosome 21q Partial Deletion Syndrome (aka: 21q22 Deletion), not including small deletions of only 21q22.3

Chromosome 22 Ring (aka: Ring 22; Ring 22, Chromosome; r22)

Chromosome 22, Trisomy Mosaic (aka: Trisomy 22 Mosaic; Trisomy 22 Mosaicism Syndrome)

Chromosome 22q11.2 duplication

Chromosome 2q32 deletion

Chromosome 2q37 deletion syndrome

Chromosome 3, Monosomy 3p2 (aka: Chromosome 3, Deletion of Distal 3p; Chromosome 3, Distal 3p Monosomy; Monosomy 3p2; Partial Deletion of Chromosome 3)

Chromosome 3, Trisomy 3q2 (aka: Chromosome 3, Distal 3q2 Duplication; Chromosome 3, Distal 3q2 Trisomy; Partial Duplication 3q Syndrome; Partial Trisomy 3q Syndrome)

Chromosome 4 Ring (aka: Ring 4; Ring 4, Chromosome; r4)

Chromosome 4, Monosomy 4q (aka: Interstitial Deletion of 4q; Proximal Deletion of

4q;Terminal Deletion of 4q; Chromosome 4Q minus micro deletion)

Chromosome 4, Monosomy Distal 4q (aka: 4q Deletion Syndrome, Partial; Chromosome 4, 4q Terminal Deletion Syndrome; Chromosome 4, Partial Monosomy 4q; Del(4q) Syndrome, Partial; Distal 4q Monosomy; Distal 4q- Syndrome; Chromosome 4Q minus micro deletion)

Chromosome 4, Partial Trisomy Distal 4q (aka: Chromosome 4, Partial Trisomy 4q (4q2 and 4q3); Chromosome 4, Partial Trisomy 4q (4q21-qter to 4q32-qter), Distal 4q Trisomy, Dup(4q) Syndrome; Partial Duplication 4q Syndrome; Partial Trisomy 4q Syndrome; Duplication of 4q21.21 to 31.22 and 2q34; Trisomy 4q Syndrome)

Chromosome 4, Trisomy 4p (aka: Chromosome 4 (Partial Trisomy 4p); Dup(4p) Syndrome; Duplication 4p Syndrome)

Chromosome 5, Trisomy 5p (aka: Chromosome 5, Trisomy 5p, (Complete (5p11-ter), Included); Chromosome 5, (Trisomy 5p, Partial, Included); Dup(5p) Syndrome; Duplication 5p Syndrome)

Chromosome 6 Ring (aka: Ring 6; Ring 6, Chromosome; r6)

Chromosome 6, Partial Trisomy 6q (aka: 6q+ Syndrome, Partial; Chromosome 6, Trisomy 6q2; Distal Duplication 6q; Distal Trisomy 6q; Duplication 6q, Partial; Trisomy 6q Syndrome, Partial; Trisomy 6q, Partial)

Chromosome 6p, Partial Monosomy (aka Partial Deletion of Chromosome 6p)

Chromosome 7, Partial Monosomy 7p (aka: Chromosome 7, 7p Deletion Syndrome, Partial; Chromosome 7, Partial Deletion of Short Arm; Del(7p) Syndrome, Partial; Interstitial 7p Monosomy; Partial 7p Monosomy; Terminal 7p Monosomy; Terminal 7p Monosomy)

Chromosome 7p Partial Duplication Syndrome (aka: 7p Duplication Syndrome)

Chromosome 7q duplication (aka: 7q22.3-7q36.1 duplication)

Chromosome 7q Partial Monosomy (aka: Chromosome 7q Deletion; Chromosome 7q Partial Deletion; Chromosome 7q11.22 deletion; Chromosome 7q34-36.1 deletion; Monosomy Chromosome 7q; Partial Monosomy Chromosome 7q; Monosomy Chromosome 7q34-36.1)

Chromosome 8, Monosomy 8p2 (aka: 8p- Syndrome, Partial; Chromosome 8, 8p Deletion Syndrome, Partial Chromosome 8, Partial Deletion of Short Arm; Chromosome 8, Partial Monosomy 8p2; Del(8p) Syndrome, Partial; Distal 8p Monosomy; Partial 8p Monosomy; Terminal 8p- Syndrome (8p21 to 8p23-pter))

Chromosome 8p inverted duplication/deletion syndrome

Chromosome 9 Ring (aka: r9; Ring 9; Ring 9, Chromosome)

Chromosome 9 Trisomy (aka: Complete Trisomy 9P; Partial Trisomy 9; Chromosome 9, Partial Trisomy 9P; Trisomy 9P Syndrome (Partial); Rethore Syndrome (obsolete); Duplication 9p Syndrome; Dup(9p) Syndrome; Chromosome 9, Trisomy 9pter-q11-13; Chromosome 9, Trisomy 9pter-q22-32)

Chromosome 9, Tetrasomy 9p (aka: Chromosome 9, Tetrasomy 9p Mosaicism; Mosaic Tetrasomy 9p; Tetrasomy 9p; Tetrasomy, Short Arm of Chromosome 9)

Chromosome 9, Trisomy Mosaic (aka: Trisomy 9 Mosaic; Trisomy 9 Mosaicism; Trisomy 9 Mosaicism Syndrome)

Chromosome 9q Partial Monosomy

Chromosome Xp deletion

Chromosome Xq26.2 duplication (aka: duplication of the distal portion of the long arm of the X chromosome; chromosome X duplication; chromosome Xq duplication)

Closed Head Injury with neuroradiological evidence of intracranial injury (e.g., subarachnoid hemorrhage, or intracranial hemorrhage, or swelling) (aka: Traumatic Brain Injury; TBI)

Cockayne Syndrome (aka: CS; Deafness-Dwarfism-Retinal Atrophy; Dwarfism with Renal Atrophy and Deafness; Neill-Dingwall Syndrome; Progeroid Nanism)

Coffin-Lowry syndrome (aka: Coffin Syndrome; Mental Retardation with Osteocartilaginous Abnormalities)

Coffin-Siris syndrome (aka: Dwarfism-Onychodysplasia; Fifth Digit Syndrome; Mental Retardation with Hypoplastic 5th Fingernails and Toenails; Short Stature-Onychodysplasia)

Cohen Syndrome (aka: Pepper Syndrome)

Colpocephaly

Congenital or acquired absence of limb (aka: severe limb deficiency; severe deficiency of extremity; severe form of Fibular Hemimelia; bilateral foot amputations at the ankle)

Connexin 26 gene mutation (with mutation on both copies and a diagnosed unilateral or bilateral hearing loss)

Cornelia de Lange Syndrome (aka: BDLS; Brachmann-de Lange Syndrome; CdLS; de Lange Syndrome)

Cortical Dysplasia

Costello Syndrome (aka: FCS Syndrome; Faciocutaneoskeletal Syndrome)

Cytomegalovirus (aka: CMV; Cytomegalic Inclusion Disease; Giant Cell Inclusion Disease (CID); Human Cytomegalovirus Infection; Salivary Gland Disease, CMV Type)

- D - Established Condition

Dandy Walker Syndrome (aka: Dandy-Walker Cyst; Dandy-Walker Deformity; Hydrocephalus, Internal, Dandy-Walker Type; Hydrocephalus, Noncommunicating, Dandy-Walker Type; DWM; Luschka-Magendie Foramina Atresia)

De Barsy Syndrome (aka: Corneal Clouding-Cutis Laxa-Mental Retardation, Cutis Laxa-Growth Deficiency Syndrome; De Barsy-Moens-Diercks Syndrome; Progeroid Syndrome of De Barsy)

De Sanctis Cacchione Syndrome (aka: Xerodermic Idiocy)

Dejerine Sottas Disease (aka: Hereditary Motor Sensory Neuropathy Type III; HSMN

Type III; Hypertrophic Interstitial Neuritis; Hypertrophic Interstitial Neuropathy; Hypertrophic Interstitial Radiculoneuropathy, Onion-Bulb Neuropathy)

Deletion 18p Syndrome (aka: Chromosome 18, Monosomy 18p; 18p Deletion Syndrome; 18p- Syndrome; Del(18p) Syndrome; Monosomy 18p Syndrome; Short Arm 18 Deletion Syndrome; Partial Deletion of Chromosome 18)

Deletion 18q syndrome (aka: Chromosome 18q- Syndrome; 18q Deletion Syndrome; 18q- Syndrome; Chromosome 18 Long Arm Deletion Syndrome; Chromosome 18, Monosomy 18Q; Del(18q) Syndrome; Monosomy 18q Syndrome; Partial Deletion of Chromosome 18)

Deletion 5p Syndrome (aka: Cri du chat Syndrome; Chromosome 5p-syndrome; Cat's Cry Syndrome; Chromosome 5, Monosomy 5p; Chromosome 5p-Syndrome; Le Jeune Syndrome; Partial Deletion of the Short Arm of Chromosome 5 Syndrome)

Deletion 9p Syndrome (aka: Chromosome 9, Partial Monosomy 9p; 9p Partial Monosomy; 9p- Syndrome, Partial; Chromosome 9, Partial Monosomy 9p22; Chromosome 9, Partial; Monosomy 9p22-pter; Del(9p) Syndrome, Partial; Deletion 9p Syndrome, Partial; Distal 9p- Syndrome; Distal Monosomy 9p; Monosomy 9p, Partial; Partial Deletion of Short Arm of Chromosome 9; 9p Deletion; 9p Minus Syndrome)

Depression of Infant and Early Childhood: Type I - Major Depression (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Deprivation/Maltreatment Disorder (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Diastrophic Dysplasia (aka: DD; DTD; Diastrophic Dwarfism; Diastrophic Nanism Syndrome)

Diencephalic Syndrome (aka: Diencephalic Syndrome of Childhood; Diencephalic Syndrome of Emaciation; Paramedian Diencephalic Syndrome; Russell's Diencephalic Cachexia; Russell's Syndrome)

DOOR Syndrome (aka: DOOR(S) Syndrome; Deafness, Onychodystrophy, Osteodystrophy,and Mental Retardation)

Down Syndrome (aka: Trisomy 21 Syndrome; Chromosome 21, Mosaic 21 Syndrome; Chromosome 21, Translocation 21 Syndrome; Trisomy G Syndrome)

Dravet Syndrome (aka: severe myoclonic epilepsy of infancy; SMEI)

Dubowitz Syndrome (aka: Intrauterine Dwarfism)

Duchenne Muscular Dystrophy (aka: Childhood Muscular Dystrophy; DMD; Muscular

Dystrophy (Classic X-linked Recessive); Progressive Muscular Dystrophy of Childhood; Pseudohypertrophic Muscular Dystrophy)

Duplication 10q Syndrome (aka: Chromosome 10, distal trisomy 10q; Chromosome 10, Partial Trisomy 10q24-qter; Chromosome 10, Trisomy 10q2; Distal Duplication 10q; Distal Trisomy 10q Syndrome; Dup(10q) Syndrome)

Dyggve Melchior Clausen Syndrome (aka: DMC Disease; DMC Syndrome; Smith-McCort Dysplasia)

Dystonia Musculorum Deformans (aka: Torsion Dystonia)

- E - Established Condition

Emanuel Syndrome (aka: Emanual Syndrome; Derivative 22; der(22) chromosome; Supernumerary der(22) Syndrome)

Encephalitis, Herpes Simplex (aka: HSE; Herpes Encephalitis; Herpetic Brainstem Encephalitis; Herpetic Meningoencephalitis)

Encephalitis, Rasmussen's (aka: Chronic Encephalitis and Epilepsy; Chronic Localized (Focal) Encephalitis; Epilepsy, Hemiplegia and Mental Retardation; Rasmussen's Syndrome)

Encephalocele (aka: Bifid Cranium, Cephalocele, Cranial Meningoencephalocele, Craniocele, Cranium Bifidum)

Encephalopathy Congenital (aka: neonatal encephalopathy)

Encephalopathy, Hypoxic Ischemic (aka: HIE; Anoxic Encephalopathy; Subacute Hypoxic Injury; Hypoxic Ischemic Brain Injury)

Encephalopathy, Static

Epidermal Nevus Syndrome (aka: Ichthyosis Hystrix Gravior; Inflammatory Linear Nevus Sebaceous Syndrome; Lambert Type Ichthyosis; Linear Nevus Sebacous Syndrome; Linear Sebaceous Nevus Sequence; Linear Sebaceous Nevus Syndrome; Nevus Sebaceous of Jadassohn; Sebaceous Nevus Syndrome)

Epilepsy

- F - Established Condition

Fahr's Disease (aka: Cerebrovascular Ferrocalcinosis; Fahr Disease; Idiopathic Basal Ganglia Calcificationl; IBGC; Nonarteriosclerotic Cerebral Calcifications; SPD Calcinosis; Striopallidodentate Calcinosis)

Familial Mental Retardation Syndromes (incl: Familial Mental Retardation Syndrome ATR-16)

Farber Disease (aka: Farber's lipogranulomatosis; ceramidase deficiency)

Feeding Disorder Associated with Concurrent Medical Condition (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker,

or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Feeding Disorder Associated with Insults to the Gastrointestinal Tract (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Feeding Disorder of Caregiver-Infant Reciprocity (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Feeding Disorder of State Regulation (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Fetal Alcohol Syndrome (aka: FAS)

Fetal Hydantoin Syndrome (aka: Dilantin Embryopathy, Phenytoin Embryopathy)

FG Syndrome (aka: Opitz-Kaveggia Syndrome)

Fiber Type Disproportion, Congenital (aka: Atrophy of Type I Fibers; CFTD; CFTDM; Myopathy of Congenital Fiber Type Disproportion; Myopathy, Congenital, With Fiber-Type Disproportion)

Fibrodysplasia Ossificans Progressiva (aka: FOP; Myositis Ossificans Progressiva)

Filippi Syndrome (aka: Syndactyly Type I with Microcephaly and Mental Retardation)

Floating Harbor Syndrome (aka: FHS; Pelletier-Leisti syndrome)

Fountain Syndrome (aka: Mental Retardation-Deafness-Skeletal Abnormalities-Coarse Face with Full Lips)

Fragile X Syndrome (aka: Fragile Site, Folic Acid Type, Rare; Fra(X)(Q27.3); Fragile X Mental Retardation Protein; FMRP; Fragile X Mental Retardation Syndrome; Marker X Syndrome; Martin-Bell Syndrome; Mental Retardation, X-Linked, Associated With Mar Xq28)

Friedreich's ataxia

Fryns Syndrome (aka: FRNS)

Fucosidosis (aka: Alpha-L-Fucosidase Deficiency)

Fukuyama Type Congenital Muscular Dystrophy (aka: Cerebromuscular Dystrophy, Fukuyama Type; Congenital Muscular Dystrophy, Fukuyama Type; FCMD; Micropolygyria With Muscular Dystrophy; Muscular Dystrophy, Congenital Progressive with Mental Retardation; Muscular Dystrophy, Congenital With Central Nervous System Involvement; Muscular Dystrophy, Congenital, Fukuyama Type; Muscular Dystrophy, Fukuyama Type)

- G - Established Condition

Galactosemia (aka: Classic Galactosemia; GALT Deficiency; Galactose-1-Phosphate Uridyl Transferase Deficiency)

Gaucher Disease Type III (Subacute/Chronic form, or "Atypical, due to Saposin C Deficiency") (aka: Gaucher Disease, Cardiovascular Form; Gaucher Disease, Type 3)

Gaucher Disease, Type II ("Perinatal Lethal" or Colloidal Type) (aka: Gaucher Disease, Type 2; Glucocerebrosidase Deficiency; Glucosylceramidase Deficiency)

Generalized Anxiety Disorder (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Glycinemia (aka: glycine encephalopathy; nonketotic hyperglycinemia; transient neonatal hyperglycinemia)

Gm1 Gangliosidosis (aka: Beta-Galactocidase-1 Deficiency; GLB1 Deficiency; Galactocidase, Beta-1; GLB1; Morquio Disease, Type B; Elastin-Binding Protein)

Griscelli Syndrome, type 1

- H - Established Condition

Hallervorden-Spatz Disease (aka: Pantothenate kinase-associated neurodegeneration; PKAN)

Hallgren Syndrome

Hearing Loss (diagnosis with bilateral hearing loss)

Hemimegalencephaly

HIV (where the child's status of the HIV infecton has been confirmed)

Holoporencephaly (aka: Alobar Holoprosencephaly; Arhinencephaly; Familial Alobar Holoprosencephaly; HS; Holoprosencephaly Malformation Complex; Holoprosencephaly Sequence; Lobar Holoprosencephaly; Semilobar Holoprosencephaly)

Homocystinuria

Human HOXA1 Syndromes (aka: Athabaskan Brainstem Dysgenesis Syndrome (ABDS); Navajo Brainstem Syndrome; Bosley-Salih-Alorainy Syndrome; BSAS)

Hunter Syndrome (aka: MPSII, MPS Disorder II, Mucopolysaccharidosis Type II)

Hurler Syndrome (aka: Mucopolysaccharidosis Type I; MPS I-H; MPS1)

Hydranencephaly

Hydrocephalus, Congenital (aka: Benign Hydrocephalus, Communicating Hydrocephalus, Internal Hydrocephalus, Non-Communicating Hydrocephalus, Normal Pressure Hydrocephalus, Obstructive Hydrocephalus)

Hypomelanosis of Ito (aka: Incontinentia pigmentosa acromians)

- I - Established Condition

I Cell Disease

Ichthyosis, Sjogren Larsson Syndrome

Infantile Anorexia (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Infantile Neuroaxonal Dystrophy (aka: INAD or Seitelberger Disease)

Infantile Spasms, Epilepsy (aka: Infantile Myoclonic Seizures, Infantile Spasm; Hypsarrhythmia)

Intraventricular Hemorrhage (aka: IVH, periventricular hemorrhage, PVH) Grade III

Intraventricular Hemorrhage (aka: IVH, periventricular hemorrhage, PVH) Grade IV

IUGR (qualifies children under one year only - evidence shows the child was born at 33 weeks gestational age and weighing 1,325 grams (2 pounds, 15 ounces) or less at birth or shortly after birth) (aka: for the purposes of determining EI Colorado eligibility IUGR can also be referred to as SGA or LBW)

IUGR (qualifies children under one year only - evidence shows the child was born at 34 weeks gestational age and weighing 1,500 grams (3 pounds, 5 ounces) or less at birth or shortly after birth) (aka: for the purposes of determining EI Colorado eligibility IUGR can also be referred to as SGA or LBW)

IUGR (qualifies children under one year only - evidence shows the child was born at 35 weeks gestational age and weighing 1,700 grams (3 pounds, 12 ounces) or less at birth or shortly after birth) (aka: for the purposes of determining EI Colorado eligibility IUGR can also be referred to as SGA or LBW)

IUGR (qualifies children under one year only - evidence shows the child was born at 36 weeks gestational age and weighing 1875 grams (4 pounds, 2 ounces) or less at birth or shortly after birth) (aka: for the purposes of determining EI Colorado eligibility IUGR

can also be referred to as SGA or LBW)

IUGR (qualifies children under one year only - evidence shows the child was born between 37 and 40 weeks gestational age and weighing less than 2,000 grams (4 pounds, 5 ounces) or less at birth or shortly after birth) (for the purposes of determining EI Colorado eligibility IUGR can also be referred to as SGA or LBW)

- J - Established Condition

Jervell and Lange-Nielsen Syndrome

Johanson-Blizzard Syndrome

Joubert Syndrome

Juberg-Marsidi Syndrome

- K - Established Condition

Kabuki Make-up Syndrome

KBG Syndrome

Keratitis Ichthyosis Deafness Syndrome

Kernicterus

Klinefelter Syndrome (aka: XXY Syndrome)

Kufs Disease (aka: Jansk

Kugelberg Welander Syndrome (aka: Wohlfart-Kugelberg-Welander syndrome or mild SMA)

- L - Established Condition

L1 Syndrome

Lambert-Eaton Myasthenic Syndrome

Landau Kleffner Syndrome

Langer-Giedion Syndrome (aka: Tricho-Rhino-Phalangeal Syndrome Type II, TRP II)

Laurence Moon Syndrome (aka: Adipogenital-Retinitis Pigmentosa Syndrome; Laurence Syndrome; LM Syndrome)

LBW < 400 grams at birth or shortly after birth (qualifies children under one year only - infant born at any gestational age (for the purposes of determining eligibility for EI Colorado LBW can also be referred to as SGA or IUGR)

LBW 1,000 - 1,199 grams at birth or shortly after birth (qualifies children under one year only - infant born at any gestational age (for the purposes of determining eligibility for EI Colorado LBW can also be referred to as SGA or IUGR)

LBW 400 - 599 grams at birth or shortly after birth (qualifies children under one year only - infant born at any gestational age (for the purposes of determining eligibility for

EI Colorado LBW can also be referred to as SGA or IUGR)

LBW 600 - 799 grams at birth or shortly after birth (qualifies children under one year only - infant born at any gestational age (for the purposes of determining eligibility for EI Colorado LBW can also be referred to as SGA or IUGR)

LBW 800 - 999 grams at birth or shortly after birth (qualifies children under one year only - infant born at any gestational age (for the purposes of determining eligibility for EI Colorado LBW can also be referred to as SGA or IUGR)

Lead encephalopathy (aka: lead poisoning encephalopathy)

Leber Congenital Amaurosis

Leigh's Disease

Lennox Gastaut Syndrome

Lenz Microphthalmia Syndrome

Lesch-Nyhan syndrome (LNS)

Leukodystrophy (aka: White Matter Disease; Metachromatic Leukodystrophy; Krabbe Disease; Krabbe Leukodystrophy; globoid cell leukodystrophy; galactosylceramide lipidosis; Canavan Disease; ACY2 Deficiency; ASP Deficiency; ASPA Deficiency; Aminoacylase-2 Deficiency; Aspartoacylase Deficiency; CD; Canavan's Leukodystrophy; Canavan-Van Bogaert-Bertrand Disease; Spongy Degeneration of the Central Nervous System; Spongy Degeneration of the Neuroaxis; Van Bogaert-Bertrand Syndrome; Vanishing White Matter Syndrome)

Levy-Yeboa Syndrome

Ligase IV Syndrome (aka: Ligase IV Deficiency; LIG4 Syndrome)

Linear Sebaceous Nevus Sequence (aka: Linear Sebaceous Nevus Syndrome; Sebaceous Nevus Syndrome; Linear Epidermal Nevus Syndrome; LEN Syndrome; Jadassohn nevus phakomatosis; JNP)

Lipodystrophy, generalized (aka: Berardinelli Lipodystrophy; Berardinelli Lipodystrophy Syndrome; Congenital Generalized Lipodystrophy)

Lissencephaly

Locked In Syndrome

Lowe Syndrome (aka: Oculocerebrorenal Syndrome)

Lysosomal Storage Disorders

- M - Established Condition

Macrocephaly, Cutis Marmorata Telangiectatica Congenita Syndrome (aka: M-CMTC)

Maple Syrup Urine Disease (where the diagnosis is late, or there is no or inadequate treatment) (aka: BCKD Deficiency, Branched Chain Alpha-Ketoacid Dehydrogenase Deficiency, Branched Chain Ketonuria I, Classical Maple Syrup Urine Disease)

Marden Walker Syndrome

Marinesco Sjogren Syndrome

Marshall Smith Syndrome

Maxillofacial Dysostosis

Meckel-Gruber Syndrome (aka: Meckel Syndrome (w/ skull defect); Dysencephalia Splanchnocystica)

Megalocornea Mental Retardation Syndrome (aka: MMR Syndrome; Neuhauser Syndrome)

MELAS Syndrome

Meningitis (including but not limited to the following types: Bacterial, Infantile, Neonatal, Meningococcal, Pneumococcal, Tuberculous, Epidemic Cerebrospinal, Pyogenic; Waterhouse-Friederichsen Syndrome)

Menkes Syndrome (aka: Menkes Disease; Kinky Hair Disease)

MERRF Syndrome

MHBD Deficiency (a.k.a. 2-methyl-3-hydroxybutyryl-CoA Dehydrogenase Deficiency)

Microcephaly

Moebius Sequence

Motor Neuron Disease

Mowat-Wilson Syndrome

Mucolipidosis IV

Mucopolysaccharidosis (except for type IVB)

Mucopolysaccharidosis VII (aka: Sly disease; Sly Syndrome; beta-glucuronidase deficiency)

Multiple Sulfatase Deficiency

Multisystem Developmental Disorder (aka: MSDD) (as defined within DC:0-3R) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Myasthenia Gravis (familial infantile type)

Myhre Syndrome

Myoclonic Encephalopathy of Childhood (Kinsbourne Syndrome)

Myopathy, Congenital

Myopathy, Congenital, Batten Turner Type

Myopathy, Desmin Storage (aka: DSM)

Myopathy, Scapuloperoneal

Myotonic dystrophy type 1 (aka: DM; DM1; Curschmann-Batten-Steinert syndrome; Steinert disease; dystrophia myotonia; myotonia atrophica)

Myotubular Myopathy

- N - Established Condition

Nemaline Myopathy

Neu Laxova Syndrome

Neuropathy, Ataxia and Retinitis Pigmentosa

Neuropathy, Congenital Hypomyelination (aka: Charcot-Marie-Tooth Type 4E; CHN; CMT4E; Congenital Dysmyelinating Neuropathy; Congenital Hypomyelinating Polyneuropathy; Congenital Hypomyelination; Congenital Hypomyelination Neuropathy; Congenital Hypomyelination (Onion Bulb), Polyneuropathy; Congenital Neuropathy caused by Hypomyelination; Hypomyelination Neuropathy)

Neuropathy, Giant Axonal

Neuropathy, Hereditary Sensory, Type I (aka: HSAN1; HSN1; Hereditary Sensory and Autonomic Neuropathy Type 1)

Neuropathy, Hereditary Sensory, Type II

Neuropathy, Hereditary Sensory, Type IV (aka: Familial Dysautonomia, Type II; Hereditary Sensory and Autonomic Neuropathy IV; HSAN IV; HSN IV; Insensitivity to Pain, Congenital, with Anhydrosis; CIPA; Neuropathy, Congenital Sensory, with Anhydrosis)

Neuropathy, Peripheral

Niemann-Pick Disease (Classic Infantile and Juvenile)

Nonketotic Hyperglycinemia

Norrie's Syndrome (aka: Anderson-Warburg Syndrome; Atrophia Bulborum Hereditaria; Episkopi Blindness; Fetal Iritis Syndrome; ND; NDP; Norrie Syndrome; Whitnall-Norman Syndrome)

- O - Established Condition

Oculocerebral Syndrome with Hypopigmentation (aka: Cross Syndrome; Kramer Syndrome)

Oculocerebrocutaneous Syndrome (aka: Delleman Syndrome; Delleman-Oorthuys Syndrome; OCC Syndrome; OCCS; Orbital Cyst with Cerebral and Focal Dermal Malformations)

Ohtahara syndrome (aka: early infantile epileptic encephalopathy 1)

Olivopontocerebellar Atrophy, Hereditary (aka: Hereditary OPCA)

Opitz G/BBB Syndrome (aka: BBBG Syndrome; Hypertelorism with Esophageal

Abnormalities and Hypospadias; Hypertelorism-Hypospadias Syndrome; Hypospadias-Dysphagia Syndrome; Opitz BBB Syndrome; Opitz BBB/G Compound Syndrome; Opitz BBBG Syndrome; Opitz G Syndrome; Opitz Hypertelorism-Hypospadias Syndrome; Opitz Oculogenitolaryngeal Syndrome; Opitz Syndrome; Opitz Syndrome; Opitz-Frias Syndrome; Telecanthus-Hypospadias Syndrome)

Opsoclonus-Myoclonus Syndrome (aka: Dancing Eyes-Dancing Feet; Kinsbourne Syndrome; Myoclonic Encephalopathy, Kinsbourne Type; Opsoclonic Encephalopathy)

Optico-Cochleo-Dentate Degeneration

Oral-Facial-Digital Syndrome (aka: OFD Syndrome; Orofaciodigital Syndrome)

Ornithine Transcarbamylase Deficiency (aka: OTC)

Orocraniodigital Syndrome (aka: Juberg Hayward Syndrome; Cleft Lip/Palate with Abnormal Thumbs and Microcephaly; Cranio-Oro-Digital Syndrome; Digital-Oro-Cranio Syndrome)

Otopalatodigital Syndrome Type I and II (aka: Taybi Syndrome; Cranioorodigital Syndrome; FPO; Faciopalatoosseous Syndrome; OPD Syndrome)

- P - Established Condition

Pachygyria

Pallister Killian Mosaic Syndrome (aka: tetrasomy 12p; Killian/Teschler-Nicola Syndrome)

Pallister W Syndrome

Paraplegia, Hereditary Spastic

Parent Infant Relationship-Global Assessment Scale (PIR-GAS) of 40 or less (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Partial Trisomy 12

Partial-Cerebellar Ataxia-Mental Retardation

Peripheral Dysostosis-Nasal Hypoplasia-Mental Retardation (aka: PNP; Acrodysostosis)

Perisylvian Syndrome, Congenital Bilateral

Periventricular Leukomalacia (PVL)

Pervasive Developmental Disorder (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner

Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Phenylketonuria (Untreated)

Phocomelia Syndrome (aka: Roberts SC-Phocomelia Syndrome; Roberts Tetraphocomelia Syndrome; SC Phocomelia Syndrome; Pseudo-thalidomide Syndrome)

Phosphoglycerate Kinase Deficiency (aka: Anemia, Hemolytic with PGK Deficiency; Erythrocyte Phosphoglycerate Kinase Deficiency; PGK; Phosphoglycerokinase)

Pick's Disease

Polymicrogyria, bilateral (aka: Congenital Bilateral Perisylvian Syndrome)

Pompe Disease

Porencephaly

Posttraumatic Stress Disorder (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Potocki-Lupski Syndrome (aka Chromosome 17p Duplication)

Prader-Willi Syndrome

Progressive Cystic Encephalomalacia

Progressive Multifocal Leukoencephalopathy

Progressive Myoclonus Epilepsy

Pseudo Hurler Polydystrophy (aka: mucolipidosis type III)

Pyruvate Carboxylase Deficiency

Pyruvate Dehydrogenase Deficiency (PDCD)

- R - Established Condition

Recombinant Chromosome 8 Syndrome (aka: Rec8 Syndrome; San Luis Valley Syndrome)

Refsum Syndrome (aka: DOC 11 (Phytanic Acid Type); Disorder of Cornification 11 (Phytanic Acid Type) ; Heredopathia Atactica Polyneuritiformis; Hypertrophic Neuropathy of Refsum; Phytanic Acid Storage Disease)

Regulation Disorders of Sensory Processing: Hypersensitive, Type A: Fearful/Cautious (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2

day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Regulation Disorders of Sensory Processing: Hyposensitive/Underresponsive (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Regulation Disorders of Sensory Processing: Sensory Stimulation-Seeking/Impulsive (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Retinoic Acid Embryopathy (aka: Accutane Embryopathy; Accutane (Fetal Effects of); Isotretinoin Embryopathy; Isotretinoin Teratogen Syndrome; Isotretinoin (Fetal Effects of); Fetal Retinoid Syndrome)

Rett Syndrome (aka: Rett Disorder)

Rhombencephalosynapsis (aka: RES)

Roberts Syndrome

Rosenberg Chutorian Syndrome

Roussy Levy Syndrome (aka: Charcot-Marie-Tooth Disease (Variant); Charcot-Marie-Tooth-Roussy-Levy Disease; Hereditary Areflexic Dystasia; Hereditary Motor Sensory Neuropathy; Hereditary Motor Sensory Neuropathy I; HMSN I)

Rubella, Congenital

Rubinstein Taybi Syndrome (aka: Broad Thumbs and Great Toes, Characteristic Facies, and Mental Retardation; Michail-Matsoukas-Theodorou-Rubinstein-Taybi Syndrome; RSTS; Rubinstein Taybi (RTS) Broad Thumb-Hallux syndrome; Rubinstein syndrome)

- S - Established Condition

Sandhoff Disease

Sanfilippo Syndrome (aka: Mucopolysaccharidosis Type III)

Santavuori Disease (aka: CLN1; INCL; Infantile Finnish Type Neuronal Ceroid Lipofuscinosis; Balkan Disease; Infantile Neuronal Ceroid Lipofuscinosis; Infantile Type Neuronal Ceroid Lipofuscinosis; Neuronal Ceroid Lipofuscinosis Type 1; Santavuori-Haltia Disease)

Schindler Disease (aka: Alpha-N-Acetylgalactosaminidase Deficiency, Schindler Type; Alpha-NAGA Deficiency, Schindler Type; Lysosomal Alpha-N-Acetylgalactosaminidase Deficiency, Schindler Type; Alpha-Galactosidase B Deficiency; GALB Deficiency; Alpha-GalNAc Deficiency, Schindler Type;

Neuroaxonal Dystrophy, Schindler Type; Neuronal Axonal Dystrophy, Schindler Type)

Schinzel Giedion Syndrome (aka: Schinzel-Giedion Midface-Retraction Syndrome)

Schizencephaly

Schwartz Jampel Syndrome

Scott Craniodigital Syndrome

Seckel Syndrome

Separation Anxiety Disorder (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Shaken Baby Syndrome (aka: Shaken Impact Syndrome; Shaken Infant Syndrome)

SHORT Syndrome

Simpson Dysmorphia Syndrome (aka: Bulldog Syndrome; DGSX Golabi-Rosen Syndrome; Dysplasia Gigantism Syndrome, X-Linked; SDYS; SGB Syndrome; Simpson-Golabi-Behmel Syndrome)

Singleton Merten Syndrome (aka: Merten Singleton Syndrome)

Sirenomelia Sequence

Smith-Lemli-Opitz Syndrome (aka: SLO)

Smith-Magenis syndrome

Social Anxiety Disorder (Social Phobia) (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Sotos Syndrome

Specific Phobia (as defined within DC:0-3R, and diagnosed by specially-qualified professional as noted) [The person making the diagnosis must be a practicing medical or mental/behavioral health professional (i.e., physician, psychologist, psychiatrist, licensed clinical social worker, or DC:0-3R trained master's level clinician) who has taken at least the 1 1/2 to 2 day DC:0-3R Practitioner Preparation Course from a Colorado State recognized trainer; an overview or awareness level workshop does not make the person qualified.]

Spina Bifida (except for spina bifida occulta, in which the spinal cord is not exposed, but the vertebral bones aren't completely closed) (aka: meningomyelocele; myelomeningocele; MMC)

Spinal Muscular Atrophy

Spondyloepiphyseal Dysplasia, Congenital

Stroke (aka: acute neurologic syndrome; congenital stroke syndrome; neonatal stroke syndrome; brain attack; basal ganglia hemorrhage - changes in ischemia; Vein of Galen ischemia; perinatal stroke)

Sturge-Weber Syndrome

Subacute Sclerosing Panencephalitis

Succinic Semialdehyde Dehydrogenase Deficiency

Sydenham Chorea (aka: Sydenham's Chorea)

- T - Established Condition

Tay Sachs Disease (aka: Cerebromacular Degeneration; GM2 Gangliosidosis, Type 1; Hexoaminidase Alpha-Subunit Deficiency (Variant B); Infantile Cerebral Ganglioside; Infantile Sipoidosis GM-2 Gangliosideosis (Type S); Lipidosis, ganglioside, infantile; Sphingolipidosis, Tay-Sachs)

Timothy Syndrome

Transverse Myelitis (aka: Cervical Transverse Myelitis)

Triphosphate Isomerase Deficiency

Triple X syndrome (aka: Trisomy X; 47,XXX; Triplo X Syndrome)

Triploidy Syndrome (aka: triploid syndrome)

Trisomy 13 (aka: Trisomy 13 - 15; Patau Syndrome)

Trisomy 18 (aka: Edwards Syndrome)

Trisomy 8 (aka: Trisomy 8 Syndrome; Trisomy 8 mosaic; Trisomy 8 mosaicism)

TTF-1 deletion (aka NKX2 deletion)

Tuberous Sclerosis

Turner Syndrome (aka: 45X Syndrome; XO Syndrome)

- U - Established Condition

Unbalanced Chromosome Translocation

Urea Cycle Defects (where the diagnosis is late, or there is no or inadequate treatment)

- V - Established Condition

VACTERL with Hydrocephalus (aka: VACTERL Association with Hydrocephalus; VACTERL-H Association; VATER Association with Hydrocephalus; VACTERL-H Association; VATER Association with Hydrocephalus)

Van der Knapp Syndrome (aka: Megalencephalic leukoencephalopathy with subcortical cysts)

Visual Impairment, Bilateral (not correctable with treatment, surgery, glasses, or contact lenses) (aka bilateral vision impairment bilateral vision loss, bilateral blindness)

- W - Established Condition

WAGR Syndrome

Walker Warburg Syndrome (aka: WWS)

Watson Syndrome (aka: Pulmonic Stenosis with Cafe-Au-Lait Spots; Cafe-Au-Lait Spots with Pulmonic Stenosis)

Weaver Syndrome

Weill Marchesani Syndrome

Werdnig Hoffman Disease

Wernicke-Korsakoff Syndrome

West Syndrome

Wieacker Syndrome

Wiedemann Rautenstrauch Syndrome

Williams Syndrome (aka: Williams-Beuren syndrome)

Wilson's Disease

Wolf-Hirchhorn Syndrome (aka: Partial Monsomy 4p)

Wolfram Syndrome

- X - Established Condition

X-linked creatine deficiency

Xeroderma Pigmentosum

XXXXX Syndrome (aka: Penta X Syndrome)

XXYY Syndrome

XYY

XYY Syndrome

- Z - Established Condition

Zellweger syndrome (aka: Bowen Syndrome; Cerebrohepatorenal Syndrome)